G6PC gene

glucose-6-phosphatase catalytic subunit

Normal Function

The *G6PC* gene provides instructions for making an enzyme called glucose 6-phosphatase. This enzyme is found on the membrane of the endoplasmic reticulum, which is a structure inside cells that is involved in protein processing and transport. Glucose 6-phosphatase works together with the glucose 6-phosphate translocase protein (produced from the *SLC37A4* gene) to break down a type of sugar molecule called glucose 6-phosphate. The breakdown of this molecule produces the simple sugar glucose, which is the primary source of energy for most cells in the body. The glucose 6-phosphatase enzyme is expressed (active) in the liver, kidneys, and intestines, and is the main regulator of glucose production in the liver.

Health Conditions Related to Genetic Changes

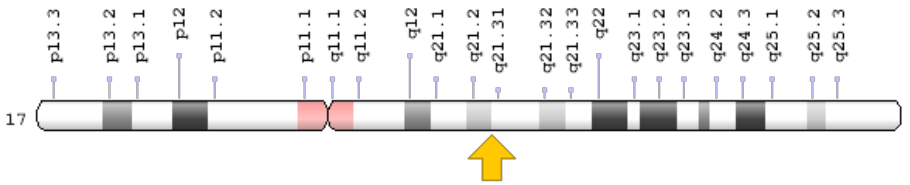
glycogen storage disease type I

At least 85 mutations in the *G6PC* gene have been found to cause glycogen storage disease type Ia (GSDIa). Most of these mutations change single protein building blocks (amino acids) in the glucose 6-phosphatase enzyme. Some specific mutations are seen more frequently in certain ethnic or racial groups. Mutations in the *G6PC* gene impair the function of the glucose 6-phosphatase enzyme. When this enzyme is not functioning normally, glucose 6-phosphate is not broken down and glucose is not produced. Glucose 6-phosphate that is not broken down to glucose is converted to fat and glycogen, a complex sugar that is stored within cells. Too much fat and glycogen stored within a cell can be toxic. This buildup damages organs and tissues throughout the body, particularly the liver and kidneys, leading to the signs and symptoms of GSDIa.

Chromosomal Location

Cytogenetic Location: 17q21.31, which is the long (q) arm of chromosome 17 at position 21.31

Molecular Location: base pairs 42,900,797 to 42,914,433 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- G-6-Pase
- G6Pase
- G6Pase-alpha
- G6PC_HUMAN
- glucose-6-phosphatase
- glucose-6-phosphatase alpha
- glucose-6-phosphatase, catalytic subunit

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Generation of Glucose from Glucose 6-Phosphate (image)
  https://www.ncbi.nlm.nih.gov/books/NBK22591/figure/A2279/
- Biochemistry (fifth edition, 2002): Liver Contains Glucose 6-phosphatase, a Hydrolytic Enzyme Absent from Muscle
  https://www.ncbi.nlm.nih.gov/books/NBK22467/#A2923

GeneReviews

- Glycogen Storage Disease Type I
  https://www.ncbi.nlm.nih.gov/books/NBK1312

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28G6PC%5BTIAB%5D%29+OR+%28%28G6Pase%5BTIAB%5D%29+OR+%28glucose-6-phosphatase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- GLUCOSE-6-PHOSPHATASE, CATALYTIC
  http://omim.org/entry/613742

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_G6PC.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=G6PC%5Bgene%5D
- HGNC Gene Family: Glucose 6-phosphatases, catalytic
  http://www.genenames.org/cgi-bin/genefamilies/set/1074
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4056
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2538
- UniProt
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